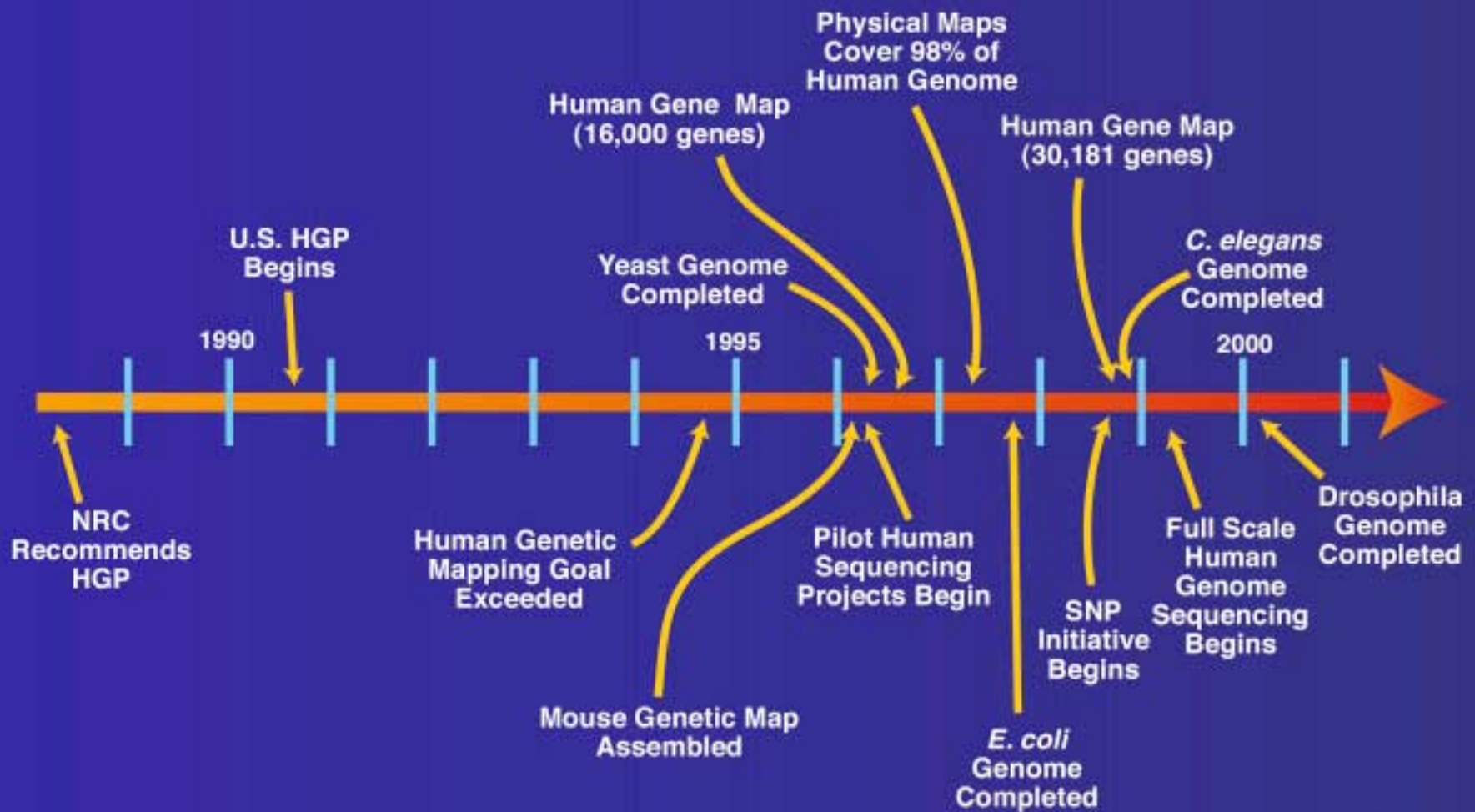


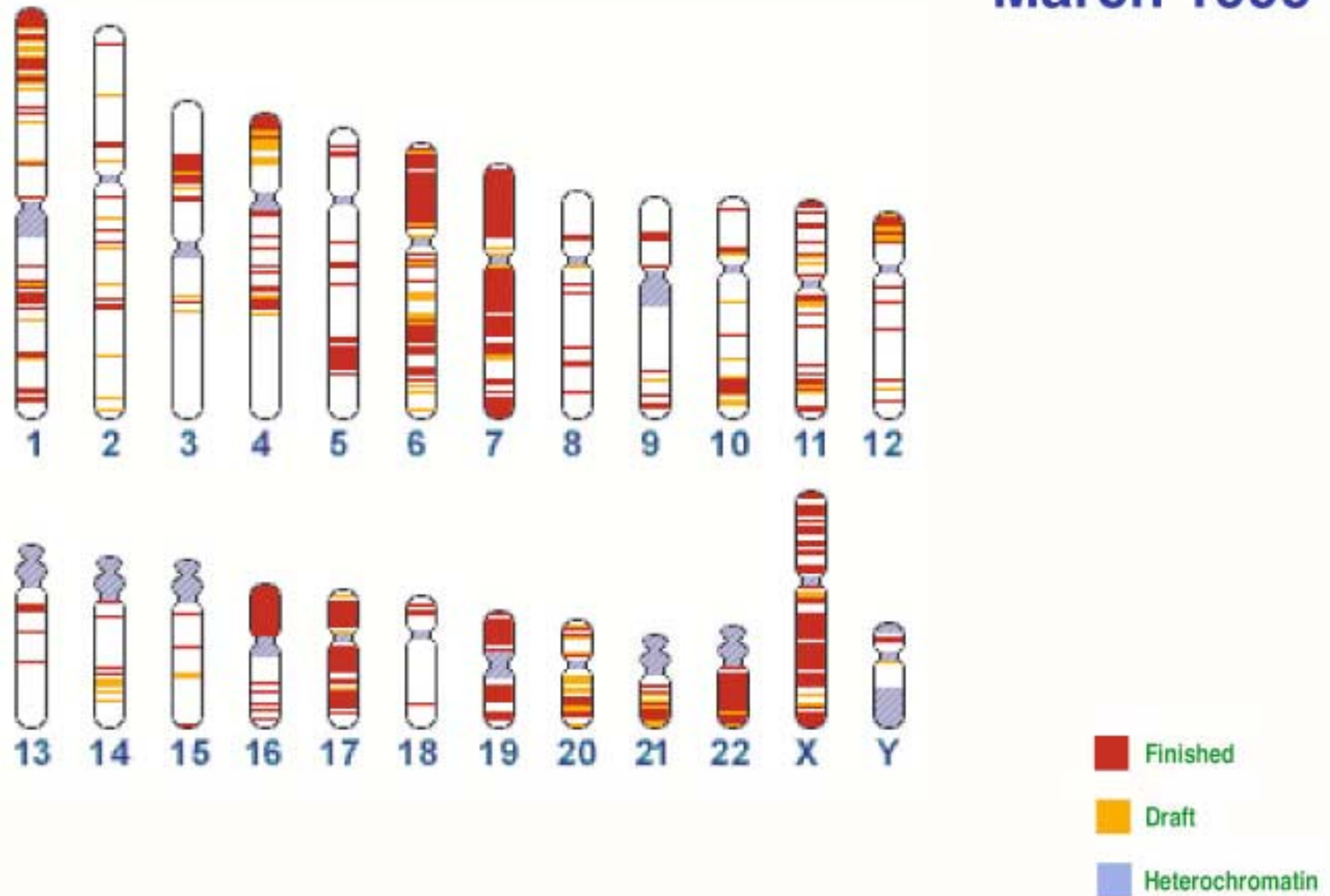
Acquiring a map, a sequence, and an increased understanding of the human genome merits a special effort that should be organized and funded specifically for this purpose... The ultimate goal would be to obtain the complete nucleotide sequence of the human genome, starting from the materials in the ordered DNA clone collection.

**Mapping and Sequencing the Human Genome
NRC Report, 1988**

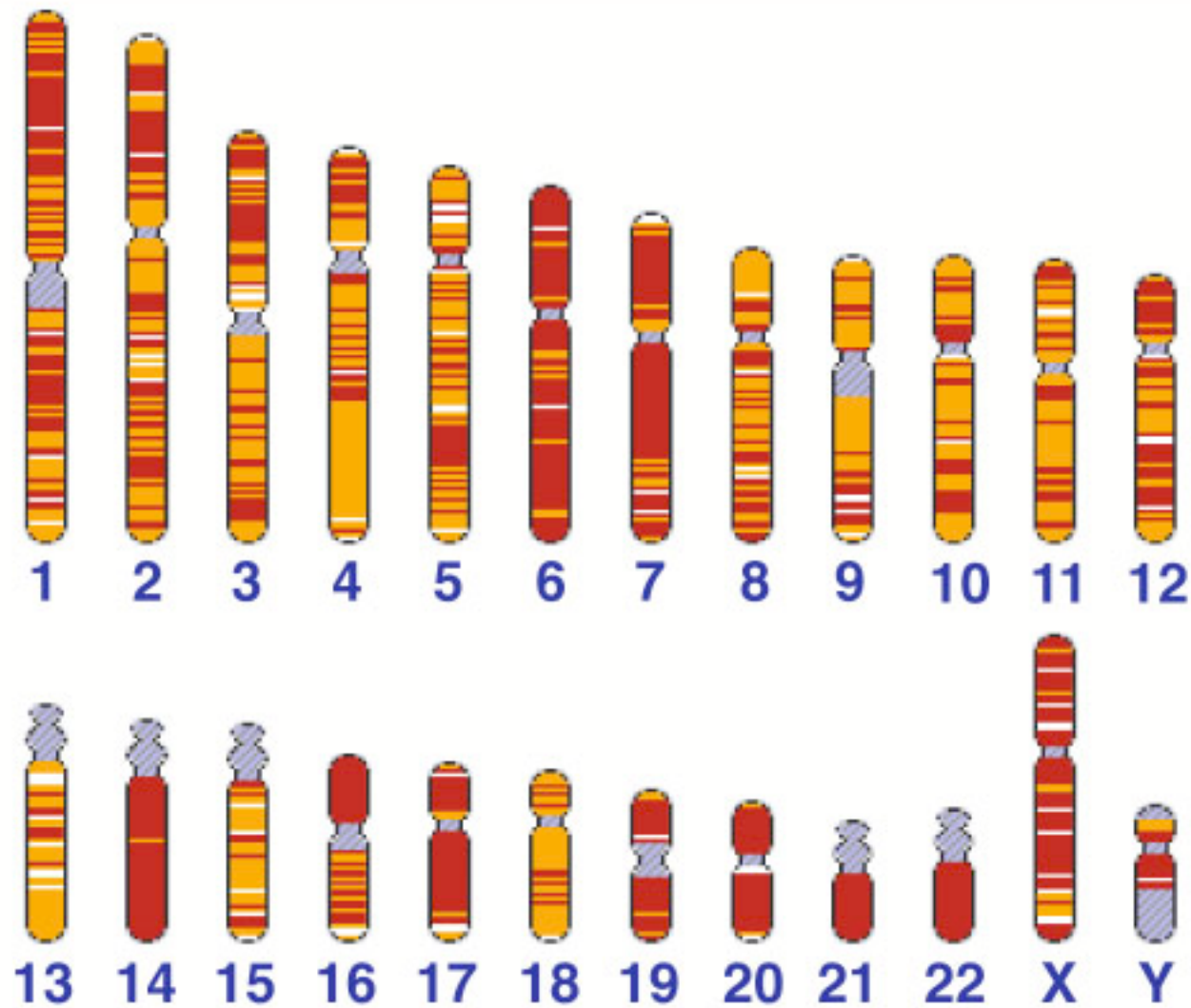




March 1999



June 2000



Finished
Draft
Heterochromatin

A Sample of Disease Genes Whose Discovery Was Aided by Access to Working Draft or Finished Sequence

Cancer:

BRCA2

X-linked Lymphoproliferative Disease

Carney Complex

Developmental Defects:

Peroxisome Biogenesis Disorder

X-linked Spondyloepiphyseal Dysplasia Tarda

Progressive Pseudorheumatoid Dysplasia

Ellis - van Creveld Syndrome

Mulibrey Nanism



A Sample of Disease Genes Whose Discovery Was Aided by Access to Working Draft or Finished Sequence

Deafness:

**Pendred Syndrome
DFNA5
DFNA13
USH1C**

Other:

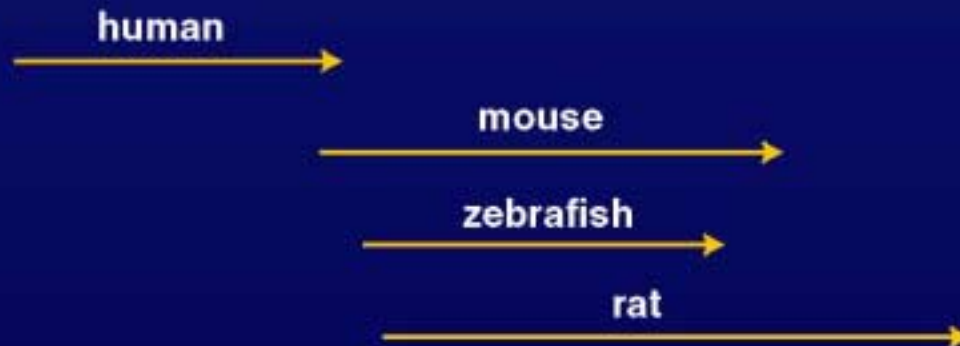
**Cerebral Cavernous Malformations
Familial Focal Segmental Glomerulosclerosis
Epilepsy with Febrile Seizures
Familial Hyperlysinemia
Hereditary Motor and Sensory Neuropathy - Lom
Total Colorblindness
May-Hegglin Anomaly**



Sequencing additional vertebrates: In series?



Or in parallel?



BUILDING THE SNP CATALOG

<u>Date</u>	<u>SNPs in Database</u>
October 1998	0
January 2000	26,274
May 2000	135,562
September 2000	1,463,574
January 2001	~3,000,000



Working draft is good
Finished sequence is better

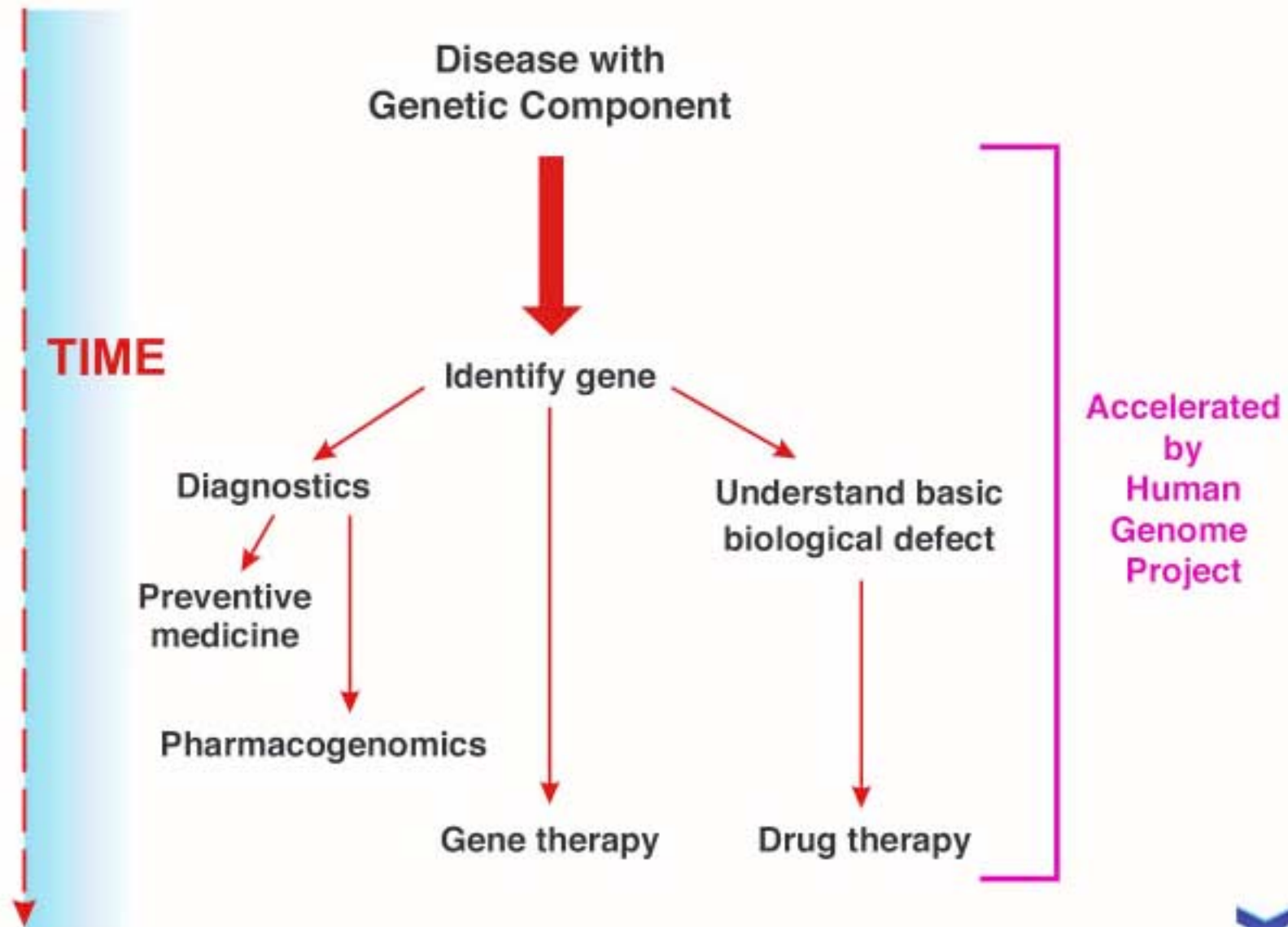


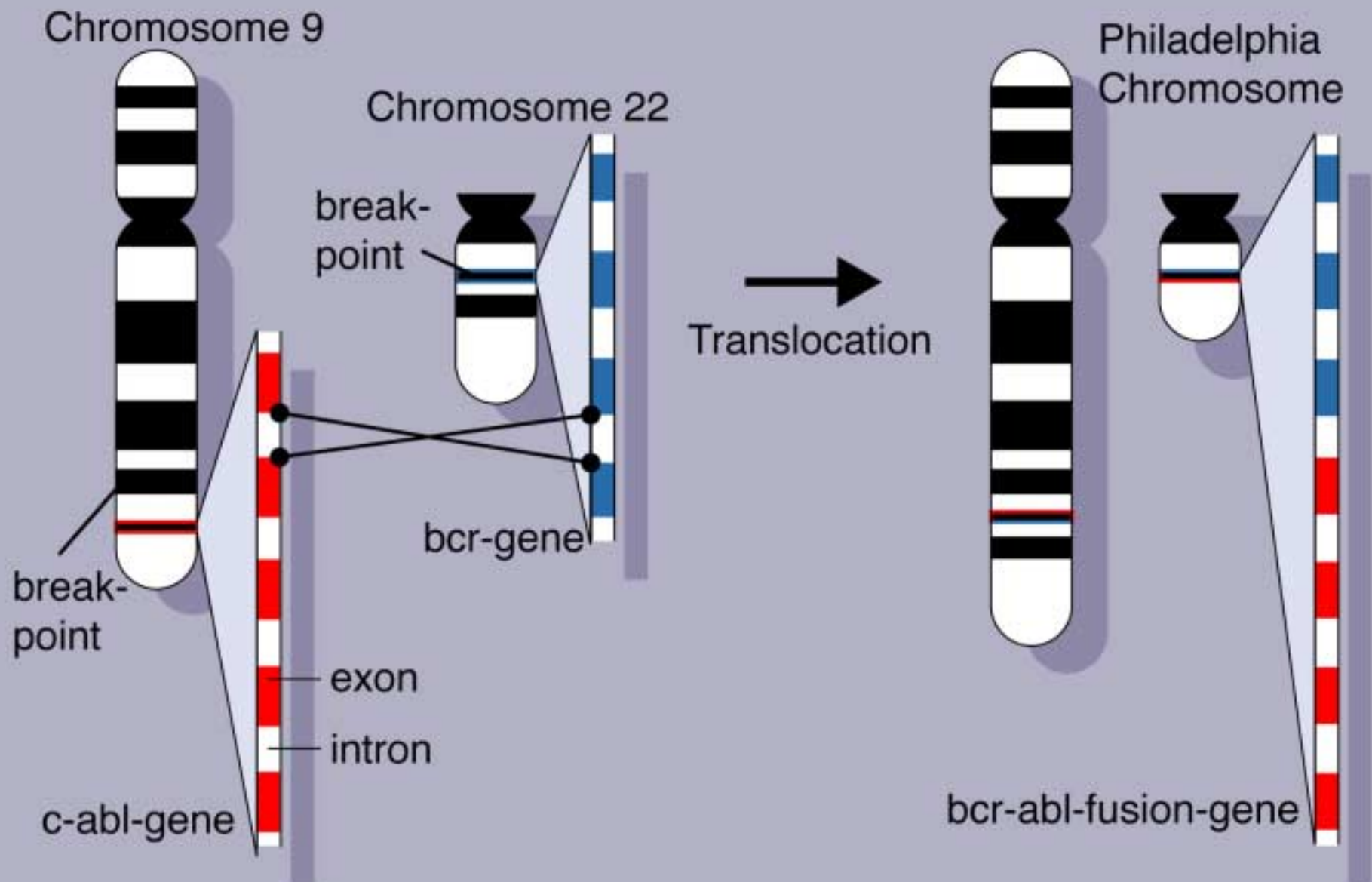


**We must never cease from exploration. And
the end of all our exploring will be to arrive
where we began and to know the place for
the first time.**

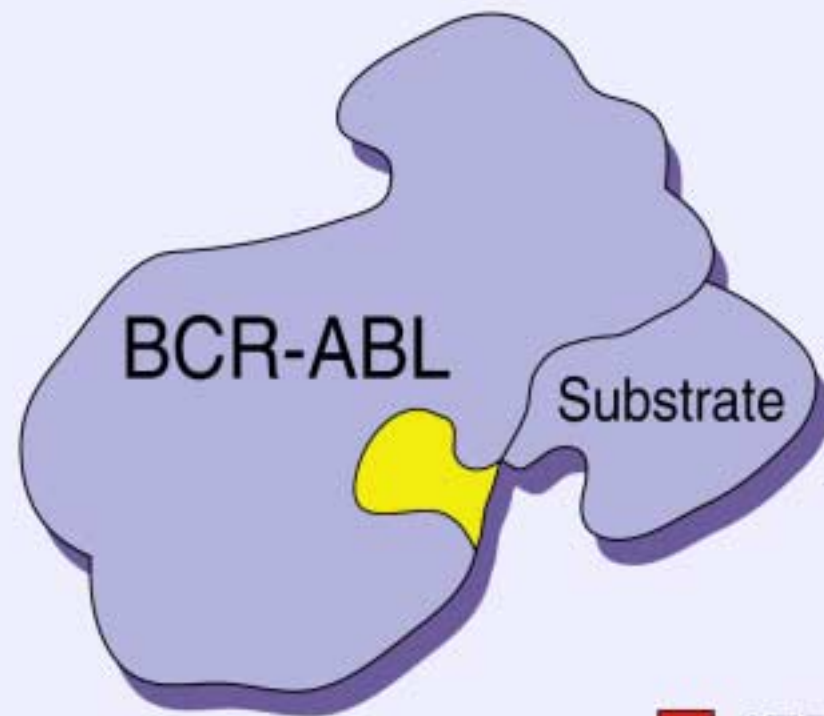
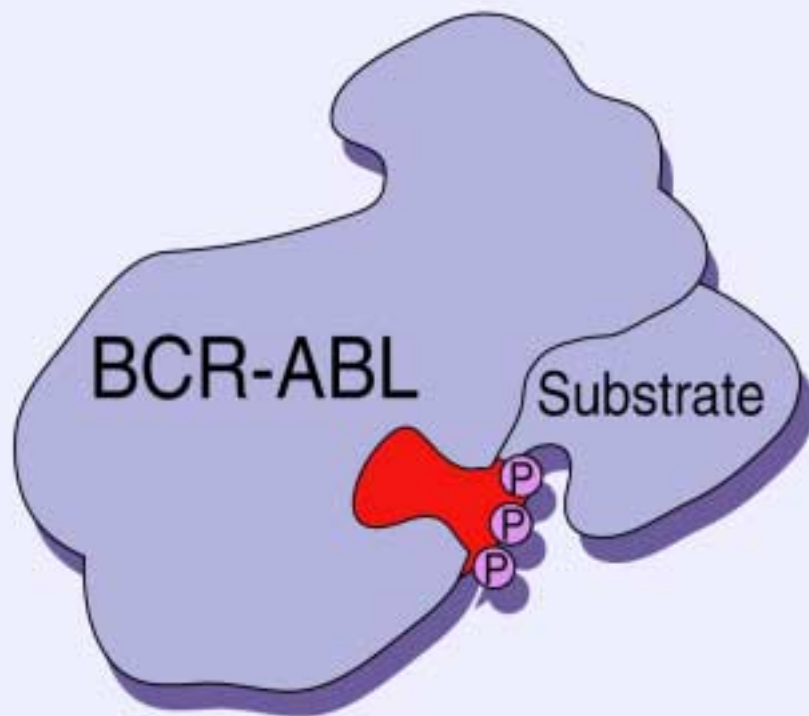
T. S. Eliot







Action of STI 571



■ ATP
■ STI571

2010

- Predictive genetic tests available for 10 conditions
- Interventions to reduce risk available for several of these
- Gene therapy successful for a few conditions
- Many primary care providers begin to practice genetic medicine
- Preimplantation diagnosis widely available, limits being fiercely debated
- Effective legislative solutions to genetic discrimination and privacy in place in US
- Access remains inequitable, especially in developing world



2020

- Gene-based designer drugs for diabetes, hypertension, etc., coming on the market
- Cancer therapy is precisely targeted to molecular fingerprint of tumor
- Dx/Rx pharmacogenomic approach is standard practice for many drugs
- Mental illness diagnosis transformed, new therapies arriving, societal views shifting
- Homologous recombination technology suggests germline gene therapy could be safe



2030

- Comprehensive genomics-based health care is the norm
Individualized preventive medicine available
Illnesses are detected early by molecular surveillance
Gene therapy and gene-based drug therapy available
for many diseases
- Full computer model of human cell replaces many
laboratory experiments
- Average life span reaches 90 years, stressing prior
socioeconomic norms
- Major anti-technology movements active in US,
elsewhere
- Serious debate is underway about humans possibly
"taking charge" of their own evolution

